

Suggested Follow-up for Maple Syrup Urine Disease  
Elevated Valine (VAL) and/or Elevated Leucine (LEU) + Isoleucine (ILE)

**Possible Causes:**

Elevated valine (VAL) and/or leucine (LEU) + isoleucine (ILE) are the primary markers for **maple syrup urine disease (MSUD)**. In MSUD, leucine, isoleucine, alloisoleucine, and valine (branched-chain amino acids) cannot be metabolized beyond their  $\alpha$ -ketoacid intermediates (due to a block in the shared catabolic pathway). Branched chain amino acids and branched chain ketoacids accumulate and produce severe toxicity often within the first 48 hours of life.

**Next Steps if Abnormal:**

**Potential medical emergency.** See infant as soon as possible to ascertain health status. Consult pediatric metabolic specialist and initiate diagnostic evaluation and treatment as recommended. Common diagnostic studies include plasma amino acids and urine organic acids. Presence of alloisoleucine in plasma is key finding for MSUD.

In addition, **repeat amino acid profile** on filter paper and send to the DHEC Public laboratory.

**Neonatal Presentation:**

Infant may show neurological deterioration in the first week of life. Lethargy and poor feeding followed by abnormal muscle tone, involuntary movements, seizures, and coma.

**Emergency Treatment:**

Treatment includes provision of enough nonprotein calories (concentrated dextrose infusion with appropriate electrolytes) to correct catabolic state and biochemical abnormalities if needed.

**Standard Treatment:**

LEU restricted/ILE, VAL controlled diet for life. Some persons with a less severe form of MSUD are thiamin responsive.

**Advice for Family:**

Provide basic information about MSUD. The handout, *When Baby Needs a Second Test for MSUD*, may be used for this purpose. Stress the importance of seeking immediate medical attention if the infant shows any signs of illness.

**Internet Resources:**

<http://ghr.nlm.nih.gov/condition=maplesyrupurinedisease>

<http://www.newbornscreening.info/Parents/aminoacid disorders/MSUD.html>

<https://www.acmg.net/PDFLibrary/Leucine.pdf>